

B96 4

(12) INTERNATIONAL APPLICATION PUBLISHED UNDER THE PATENT COOPERATION TREATY (PCT)

(19) World Intellectual Property Organization
International Bureau



(43) International Publication Date
15 February 2001 (15.02.2001)

PCT

(10) International Publication Number
WO 01/11083 A3

- (51) International Patent Classification⁷: C12Q 1/68 (74) Agent: CAMACHO, Jennifer, A.; Testa, Hurwitz & Thibault, LLP, High Street Tower, 125 High Street, Boston, MA 02110 (US).
- (21) International Application Number: PCT/US00/21763
- (22) International Filing Date: 9 August 2000 (09.08.2000) (81) Designated States (*national*): AU, CA, JP.
- (25) Filing Language: English (84) Designated States (*regional*): European patent (AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE).
- (26) Publication Language: English
- (30) Priority Data: 09/371,991 11 August 1999 (11.08.1999) US Published: — with international search report
- (71) Applicant: EXACT LABORATORIES, INC. [US/US]; 63 Great Road, Maynard, MA 01754 (US). (88) Date of publication of the international search report: 30 August 2001
- (72) Inventors: SHUBER, Anthony, P.; 11 Grant Street, Milford, MA 01757 (US). PIERCEALL, William; 17 Abbott Street, Wellesley, MA 02482 (US). For two-letter codes and other abbreviations, refer to the "Guidance Notes on Codes and Abbreviations" appearing at the beginning of each regular issue of the PCT Gazette.



WO 01/11083 A3

(54) Title: METHODS FOR DETECTING MUTATIONS USING PRIMER EXTENSION

(57) Abstract: Methods for detecting nucleotide deletions in biological samples are described. Methods of the invention are particularly useful for detecting deletions in regions of polynucleotide repeats. In particular, methods of the invention are useful to detect deletions at the BAT26 locus.

INTERNATIONAL SEARCH REPORT

Int .tional Application No

PCT/US 00/21763

A. CLASSIFICATION OF SUBJECT MATTER

IPC 7 C12Q1/68

According to International Patent Classification (IPC) or to both national classification and IPC

B. FIELDS SEARCHED

Minimum documentation searched (classification system followed by classification symbols)

IPC 7 C12Q

Documentation searched other than minimum documentation to the extent that such documents are included in the fields searched

Electronic data base consulted during the international search (name of data base and, where practical, search terms used)

EPO-Internal, WPI Data, PAJ, CHEM ABS Data, MEDLINE, BIOSIS, EMBASE

C. DOCUMENTS CONSIDERED TO BE RELEVANT

Category *	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
X	ANDREAS BRAUN ET AL: "Improved Analysis of Microsaellites Using Mass spectrometry" GENOMICS, vol. 46, 1997, pages 18-23, XP002162453	1-12, 22-30
Y	page 19, left-hand column, line 59 -page 19, right-hand column, line 3; figure 2	13-21
Y	US 5 741 650 A (LAPIDUS STANLEY N ET AL) 21 April 1998 (1998-04-21) cited in the application claims 1-5	13,14

	-/--	

☒ Further documents are listed in the continuation of box C.☒ Patent family members are listed in annex.

* Special categories of cited documents:

A document defining the general state of the art which is not considered to be of particular relevance

E earlier document but published on or after the international filing date

L document which may throw doubts on priority claim(s) or which is cited to establish the publication date of another citation or other special reason (as specified)

O document referring to an oral disclosure, use, exhibition or other means

P document published prior to the international filing date but later than the priority date claimed

T later document published after the international filing date or priority date and not in conflict with the application but cited to understand the principle or theory underlying the invention

X document of particular relevance; the claimed invention cannot be considered novel or cannot be considered to involve an inventive step when the document is taken alone

Y document of particular relevance; the claimed invention cannot be considered to involve an inventive step when the document is combined with one or more other such documents, such combination being obvious to a person skilled in the art.

Z document member of the same patent family

Date of the actual completion of the international search

12 March 2001

Date of mailing of the international search report

02/04/2001

Name and mailing address of the ISA

European Patent Office, P.B. 5818 Patentlaan 2
NL - 2280 HV Rijswijk
Tel. (+31-70) 340-2040, Tx. 31 651 epo nl,
Fax (+31-70) 340-3016

Authorized officer

Gabriels, J

INTERNATIONAL SEARCH REPORT

Int. Patent Application No.

PCT/US 00/21763

C. (Continuation) DOCUMENTS CONSIDERED TO BE RELEVANT

Category *	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
Y	HOANG JEAN-MARC ET AL: "BAT-26, an indicator of the replication error phenotype in colorectal cancers and cell lines." CANCER RESEARCH, vol. 57, no. 2, 1997, pages 300-303, XP002162454 ISSN: 0008-5472 page 300, right-hand column, line 35 -page 302, right-hand column, line 21	15-21
X	US 5 650 277 A (NAVOT NIR ET AL) 22 July 1997 (1997-07-22) figure 1; example 1	1-12, 22-30
X	WO 96 30545 A (APPLIED GENETICS INC) 3 October 1996 (1996-10-03) page 12, line 22 -page 47, line 3; claims 1-33	1-12, 22-30
Y	GISMONDI VIVIANA ET AL: "Characterization of 19 novel and six recurring APC mutations in Italian adenomatous polyposis patients, using two different mutation detection techniques." HUMAN MUTATION, vol. 9, no. 4, 1997, pages 370-373, XP002162567 ISSN: 1059-7794 table 2	19
E	WO 00 70096 A (EXACT LAB INC) 23 November 2000 (2000-11-23) the whole document	1-30

INTERNATIONAL SEARCH REPORT

Information on patent family members

International Application No

PCT/US 00/21763

Patent document cited in search report		Publication date	Patent family member(s)	Publication date
US 5747650	A	21-04-1998	AU 704696 B	29-04-1999
			AU 2240697 A	22-08-1997
			CA 2215263 A	07-08-1997
			EP 0817968 A	14-01-1998
			US 5952178 A	14-09-1999
			WO 9728450 A	07-08-1997
US 5650277	A	22-07-1997	IL 102382 A	04-08-1996
			IL 111268 A	09-05-1999
			AU 4770393 A	31-01-1994
			EP 0648222 A	19-04-1995
			FI 946127 A	20-02-1995
			IL 106199 A	24-09-1998
			IL 111267 A	14-07-1999
			JP 8507198 T	06-08-1996
			NO 945100 A	22-02-1995
			WO 9401447 A	20-01-1994
			US 5710028 A	20-01-1998
WO 9630545	A	03-10-1996	AU 5296496 A	16-10-1996
			US 5976798 A	02-11-1999
WO 0070096	A	23-11-2000	AU 5027400 A	05-12-2000